TGFB3 gene

transforming growth factor beta 3

Normal Function

The *TGFB3* gene provides instructions for producing a protein called transforming growth factor beta-3 (TGFβ-3). This protein is found throughout the body and is required for development before birth and throughout life. To carry out its functions, TGFβ-3 attaches (binds) to receptor proteins on the surface of cells. This binding triggers the transmission of signals within the cell, controlling various cellular activities. As part of a signaling pathway, called the TGF-β pathway, the TGFβ-3 protein helps control the growth and division (proliferation) of cells, the process by which cells mature to carry out specific functions (differentiation), cell movement (motility), and controlled cell death (apoptosis). Because the TGFβ-3 protein keeps cells from growing and dividing too rapidly or in an uncontrolled way, it can suppress the formation of tumors.

The TGFβ-3 protein is especially abundant in tissues that develop into the muscles used for movement (skeletal muscles), and plays a key role in their development. The protein is also involved in the formation of blood vessels, regulation of bone growth, wound healing, and immune system function.

Health Conditions Related to Genetic Changes

arrhythmogenic right ventricular cardiomyopathy

Loeys-Dietz syndrome

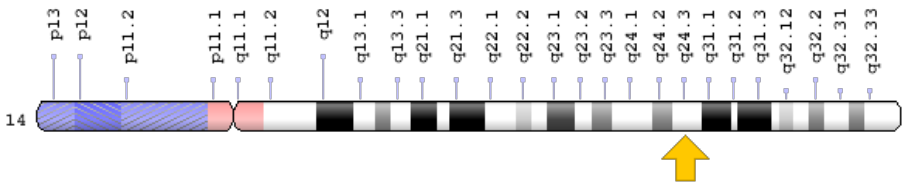
At least 11 mutations in the *TGFB3* gene have been found to cause Loeys-Dietz syndrome type V. This disorder affects connective tissue, which gives structure and support to blood vessels, the skeleton, and many other parts of the body. Loeys-Dietz syndrome type V is characterized by blood vessel abnormalities, heart defects, and skeletal deformities. The *TGFB3* gene mutations that cause this condition lead to the production of a TGFβ-3 protein with little or no function. As a result, the protein cannot bind to its receptors. Although the TGFβ-3 protein and its receptors are not bound, TGF-β pathway signaling occurs at an even greater intensity than normal. Researchers speculate that the activity of other proteins in this signaling pathway is increased to compensate for the reduction in TGFβ-3 activity; however, the exact mechanism responsible for the increase in signaling is unclear. The overactive

signaling pathway disrupts development of connective tissue and various body systems and leads to the signs and symptoms of Loeys-Dietz syndrome type V.

Chromosomal Location

Cytogenetic Location: 14q24.3, which is the long (q) arm of chromosome 14 at position 24.3

Molecular Location: base pairs 75,958,097 to 75,982,022 on chromosome 14 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- RNHF
- TGF beta 3
- TGF-beta3

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Signal Proteins of the TGF-β Superfamily Act Through Receptor Serine/Threonine Kinases and Smads https://www.ncbi.nlm.nih.gov/books/NBK26822/#A2874
- Molecular Cell Biology (fourth edition, 2000): TGFβ signaling (image) https://www.ncbi.nlm.nih.gov/books/NBK21526/figure/A7151/

GeneReviews

- Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy https://www.ncbi.nlm.nih.gov/books/NBK1131
- Loeys-Dietz Syndrome https://www.ncbi.nlm.nih.gov/books/NBK1133

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TGFB3%5BTIAB%5D%29+OR+%28transforming+growth+factor+beta+3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- TRANSFORMING GROWTH FACTOR, BETA-3
  http://omim.org/entry/190230

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_TGFB3.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=TGFB3%5Bgene%5D
- HGNC Gene Family: Endogenous ligands
  http://www.genenames.org/cgi-bin/genefamilies/set/542
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11769
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/7043
- UniProt
  http://www.uniprot.org/uniprot/P10600

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https://ghr.nlm.nih.gov/gene/TGFB3

Reviewed: March 2017
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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